

CLAIMS

- 1 A method for the diagnosis of a single nucleotide polymorphism in a α_4 integrin subunit in a human, which method comprises determining the sequence of nucleic acid of the human at one or more of the following positions:
- 5 positions 740, 2273, 2446, 3311 and 3506 in the coding region of α_4 integrin subunit gene as defined by the positions in EMBL ACCESSION NO. L12002;
position 967 in the promoter region of α_4 integrin subunit gene as defined by the position in EMBL ACCESSION NO. L26509; and
positions 184, 238, 331, 436, 676, 1010, or 1115 in the promoter region of α_4 integrin subunit
- 10 gene as defined by the position in EMBL ACCESSION NO. M26841;
and determining the status of the human by reference to polymorphism in the α_4 integrin subunit gene.
- 2 A method for diagnosis according to claim 1 in which the single nucleotide polymorphisms are further defined as:
- 15 the single nucleotide polymorphism at position 740 is presence of C and/or T;
the single nucleotide polymorphism at position 2273 is presence of A and/or G;
the single nucleotide polymorphism at position 2446 is presence of C and/or T;
the single nucleotide polymorphism at position 3311 is presence of T and/or C;
the single nucleotide polymorphism at position 3506 is presence of C and/or T;
- 20 the single nucleotide polymorphism at position 967 is presence of G and/or A;
the single nucleotide polymorphism at position 184 is presence of A and/or G;
the single nucleotide polymorphism at position 238 is presence of C and/or T;
the single nucleotide polymorphism at position 331 is presence of C and/or T;
the single nucleotide polymorphism at position 436 is presence of C and/or T;
- 25 the single nucleotide polymorphism at position 676 is presence of C and/or T;
the single nucleotide polymorphism at position 1010 is presence of C and/or A; and
the single nucleotide polymorphism at position 1115 is presence of C and/or T.
- 3 A method for diagnosis according to claim 1 in which the method comprises determining the sequence of nucleic acid of the human at position 2446 for presence of C
- 30 and/or T.

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4 A method for diagnosis according to any preceding claim in which the sequence is determined by a method selected from amplification refractory mutation system, minisequencing and restriction fragment length polymorphism.

5 A nucleic acid comprising any one of the following polymorphisms:

- 5 the nucleic acid of EMBL ACCESSION No. L12002 with T at position 740 as defined by the position in EMBL ACCESSION No. L12002;
the nucleic acid of EMBL ACCESSION No. L12002 with G at position 2273 as defined by the position in EMBL ACCESSION No. L12002;
the nucleic acid of EMBL ACCESSION No. L12002 with T at position 2446 as defined by the
10 position in EMBL ACCESSION No. L12002;
the nucleic acid of EMBL ACCESSION No. L12002 with C at position 3311 as defined by the position in EMBL ACCESSION No. L12002;
the nucleic acid of EMBL ACCESSION No. L12002 with T at position 3506 as defined by the position in EMBL ACCESSION No. L12002;
15 the nucleic acid of EMBL ACCESSION No. L26059 with A at position 967 as defined by the position in EMBL ACCESSION No. L26059;
the nucleic acid of EMBL ACCESSION No. M26841 with G at position 184 as defined by the position in EMBL ACCESSION No. M26841;
the nucleic acid of EMBL ACCESSION No. M26841 with T at position 238 as defined by the
20 position in EMBL ACCESSION No. M26841;
the nucleic acid of EMBL ACCESSION No. M26841 with T at position 331 as defined by the position in EMBL ACCESSION No. M26841;
the nucleic acid of EMBL ACCESSION No. M26841 with T at position 436 as defined by the position in EMBL ACCESSION No. M26841;
25 the nucleic acid of EMBL ACCESSION No. M26841 with T at position 676 as defined by the position in EMBL ACCESSION No. M26841;
the nucleic acid of EMBL ACCESSION No. M26841 with A at position 1010 as defined by the position in EMBL ACCESSION No. M26841;
the nucleic acid of EMBL ACCESSION No. M26841 with T at position 1115 as defined by
30 the position in EMBL ACCESSION No. M26841;
or a complementary strand thereof or an antisense sequence for a coding region or a fragment thereof of at least 20 bases comprising at least one polymorphism.

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6 A computer readable medium comprising at least one nucleic acid sequence as defined
in claim 5 stored on the medium.

7 An allele specific primer capable of detecting a α_4 integrin subunit gene polymorphism
at one or more of positions:

5 positions 740, 2273, 2446, 3311 and 3506 in the coding region of α_4 integrin subunit gene as
defined by the positions in EMBL ACCESSION NO. L12002;

position 967 in the promoter region of α_4 integrin subunit gene as defined by the position in
EMBL ACCESSION NO. L26509; and

positions 184, 238, 331, 436, 676, 1010, or 1115 in the promoter region of α_4 integrin subunit
10 gene as defined by the position in EMBL ACCESSION NO. M26841.

8 An allele-specific oligonucleotide probe capable of detecting a α_4 integrin subunit gene
polymorphism at one or more of positions:

positions 740, 2273, 2446, 3311 and 3506 in the coding region of α_4 integrin subunit gene as
defined by the positions in EMBL ACCESSION NO. L12002;

15 position 967 in the promoter region of α_4 integrin subunit gene as defined by the position in
EMBL ACCESSION NO. L26509; and

positions 184, 238, 331, 436, 676, 1010, or 1115 in the promoter region of α_4 integrin subunit
gene as defined by the position in EMBL ACCESSION NO. M26841.

9 An allelic variant of the human integrin α_4 polypeptide having a methionine at position
20 679 or a fragment thereof comprising at least 10 amino acids provided that the fragment
comprises the allelic variant at position 679.

10 Use of a α_4 integrin subunit ligand antagonist drug in preparation of a medicament for
treating a α_4 integrin subunit ligand mediated disease in a human diagnosed as having a single
nucleotide polymorphism at one or more of positions:

25 positions 740, 2273, 2446, 3311 and 3506 in the coding region of α_4 integrin subunit gene as
defined by the positions in EMBL ACCESSION NO. L12002;

position 967 in the promoter region of α_4 integrin subunit gene as defined by the position in
EMBL ACCESSION NO. L26509; and

positions 184, 238, 331, 436, 676, 1010, or 1115 in the promoter region of α_4 integrin subunit
30 gene as defined by the position in EMBL ACCESSION NO. M26841.

11 Use of a nucleic acid sequence comprising at least one of the following polymorphisms
in the promoter:

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position 967 in the promoter region of α_4 integrin subunit gene as defined by the position in EMBL ACCESSION NO. L26509; and

positions 184, 238, 331, 436, 676, 1010, or 1115 in the promoter region of α_4 integrin subunit gene as defined by the position in EMBL ACCESSION NO. M26841;

5 to identify compounds that modify expression of the human α_4 integrin subunit gene.

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